



F7 gene

coagulation factor VII

Normal Function

The *F7* gene provides instructions for making a protein called coagulation factor VII. Coagulation factors are a group of related proteins that are involved in the coagulation system, which is a series of chemical reactions that form blood clots. After an injury, clots seal off blood vessels to stop bleeding and trigger blood vessel repair.

Coagulation factor VII is made primarily by cells in the liver. The protein circulates in the bloodstream in an inactive form until the coagulation system is turned on (activated) by an injury that damages blood vessels. Activated coagulation factor VII helps turn on other coagulation factors in turn. This step-wise process ultimately promotes the conversion of an important coagulation protein called fibrinogen into fibrin, which is the material that forms blood clots.

Health Conditions Related to Genetic Changes

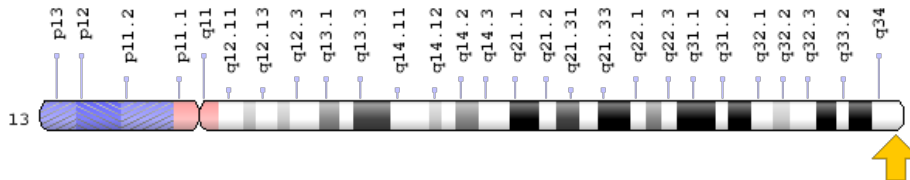
factor VII deficiency

Almost 300 mutations in the *F7* gene have been found to cause a rare bleeding disorder called factor VII deficiency. This disorder commonly causes nosebleeds, easy bruising, bleeding of the gums, and prolonged or excessive bleeding following surgery or physical injury. In severe cases, life-threatening episodes of bleeding inside the skull or gastrointestinal tract can occur. Some affected individuals have no bleeding problems. The *F7* gene mutations involved in this condition reduce the amount of coagulation factor VII in the bloodstream. A shortage of coagulation factor VII prevents blood from clotting normally, causing episodes of abnormal bleeding that can be severe. What determines the severity of the condition is unclear; it does not appear to be related to the amount of coagulation factor VII in the blood.

Chromosomal Location

Cytogenetic Location: 13q34, which is the long (q) arm of chromosome 13 at position 34

Molecular Location: base pairs 113,105,773 to 113,120,681 on chromosome 13 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FVII coagulation protein
- proconvertin
- serum prothrombin conversion accelerator
- SPCA

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Diagram: Blood-Clotting Cascade
<https://www.ncbi.nlm.nih.gov/books/NBK22589/?rendertype=figure&id=A1401>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28F7%5BTIAB%5D%29+OR+%28coagulation+factor+VII%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- COAGULATION FACTOR VII
<http://omim.org/entry/613878>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_F7.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=F7%5Bgene%5D>
- HGNC Gene Family: Gla domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/1250>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3544
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2155>
- UniProt
<http://www.uniprot.org/uniprot/P08709>

Sources for This Summary

- OMIM: COAGULATION FACTOR VII
<http://omim.org/entry/613878>
- Girolami A, Cosi E, Santarossa C, Ferrari S, Luigia Randi M. The Story of Serum Prothrombin Conversion Accelerator, Proconvertin, Stable Factor, Cothromboplastin, Prothrombin Accelerator or Autoprothrombin I, and Their Subsequent Merging into Factor VII. *Semin Thromb Hemost.* 2015 Jun;41(4):366-73. doi: 10.1055/s-0035-1549851. Epub 2015 May 14. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/25973586>
- Herrmann FH, Wulff K, Auerswald G, Schulman S, Astermark J, Batorova A, Kreuz W, Pollmann H, Ruiz-Saez A, De Bosch N, Salazar-Sanchez L; Greifswald Factor FVII Deficiency Study Group. Factor VII deficiency: clinical manifestation of 717 subjects from Europe and Latin America with mutations in the factor 7 gene. *Haemophilia.* 2009 Jan;15(1):267-80. doi: 10.1111/j.1365-2516.2008.01910.x. Epub 2008 Oct 30.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18976247>
- Mariani G, Bernardi F. Factor VII Deficiency. *Semin Thromb Hemost.* 2009 Jun;35(4):400-6. doi: 10.1055/s-0029-1225762. Epub 2009 Jul 13. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19598068>
- Mariani G, Herrmann FH, Bernardi F, Schved JF, Auerswald G, Ingerslev J. Clinical manifestations, management, and molecular genetics in congenital factor VII deficiency: the International Registry on Congenital Factor VII Deficiency (IRF7). *Blood.* 2000 Jul 1;96(1):374.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/10939805>
- Mariani G, Herrmann FH, Dolce A, Batorova A, Etro D, Peyvandi F, Wulff K, Schved JF, Auerswald G, Ingerslev J, Bernardi F; International Factor VII Deficiency Study Group. Clinical phenotypes and factor VII genotype in congenital factor VII deficiency. *Thromb Haemost.* 2005 Mar;93(3):481-7.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15735798>

Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/F7>

Reviewed: October 2016
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services